CF-AIR & CF@LANTA NEWSLETTER

Cystic Fibrosis Center of Excellence

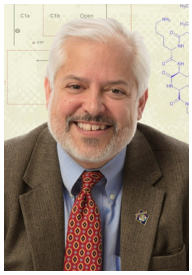
Emory University - Children's Healthcare of Atlanta - Georgia Tech - Augusta Univ. - Georgia State - UGA

NOVEMBER 2024

In This Issue

Update from Co-Directors

Drs. McCarty and Kopp



This has been a wonderful year for CF-AIR and CF@LANTA! Our efforts to expand research that bridges the bench to bedside continues to be recognized with new grants for our faculty and exciting clinical trial results. We were thrilled that our care team (pictured below) was recognized at the 38th Annual North American Cystic Fibrosis Conference (NACFC) in September for its selection as the Cystic Fibrosis Foundation's 2024 Outstanding Care Center Partnership Award. The Outstanding Partnership Award was established to recognize the care centers and their teams that have gone above and beyond in collaborating with their local CF Foundation chapters to help serve the CF community. What a tremendous honor! Hopefully, 2025 will continue to bring amazing discoveries to enable new therapies for people with CF. We thank you for your continued support!

Recent News and Updates

2024 Emory University School of Medicine Clinical Distinctions

The Clinical Distinctions (CD) designation program is designed to recognize faculty who have devoted a majority of their time and careers to providing excellence in clinical care.

Lokesh Guglani, MD
Distinguished Physician
Pediatrics

Ajay Kasi, MD
Senior Physician
Pediatrics

Global Ranking for Quorum Sensing Research

Dr. Stephen Diggle, Professor at Georgia Tech and CF-AIR investigator, has been ranked #6 globally in quorum sensing research, *according to Highly Ranked Scholars™. This ranking is based on profound impact (citations) and of utmost quality (h-index). He was also named a Top 1% Scientist in Stanford University "World's Top 2% Scientist List" in 2023 and elected to the American Academy of Microbiology. The list of top world researchers can be found online here.*

Advancing Diversity, Equity, and Inclusion

Support for Spanish-speaking Patients and Families

Josie McNeany plays a key role in helping our Spanish-speaking patients and their families feel comfortable and informed about our research. As a Clinical Research Coordinator (II) and Certified Spanish Interpreter, Josie works one-on-one with 15 patients and their families to explain what our research is about, the steps we take, and what it means for them. She also connects them with helpful resources from the Cystic Fibrosis Foundation, making sure they know what support is available. Through Josie's work and the support of the CF Bridge of Hope, a family from Honduras received the care they needed. Josie's dedication helps patients feel at ease and confident in understanding the research and their options.

Building Community and Inclusion

Emory University's Employee Resource Groups (ERGs) bring together faculty and staff to support each other and promote an inclusive, welcoming workplace. These groups offer networking, mentorship, and advocacy to strengthen Emory's commitment to diversity and inclusion.

- Emory Black Employee Network
- Emory Latinx Employee Resource Network
- Emory Pride Employee Network
- Emory Veteran Employee Network
- Emory Asian Pacific Islander Desi Employee Network
- Employees with Disabilities at Emory Network

Learn more at https://hr.emory.edu/eu/resources/ergs/index.html

Have an idea or project to help advance our DEI initiatives? Contact Clovis Sarmiento at csarmi3@emory.edu

Center Highlights

105 Primary and Affiliate CF-AIR Members

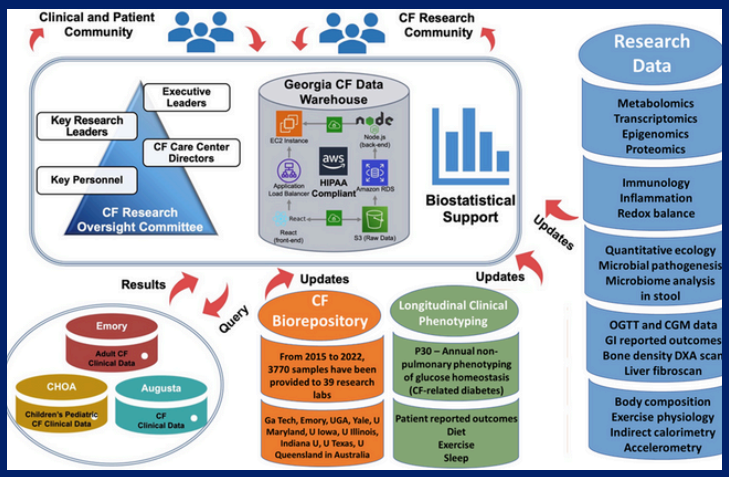
$24M awarded in Fiscal Year 2024

318 publications by 83 researchers in calendar year 2023

Ranked #2 in NIH Funding for Cystic Fibrosis

Dr. Arlene Stecenko secures NIH U24 grant to advance CF Research

Arlene Stecenko, MD, has received the Department of Pediatrics' first-ever NIH U24 grant to expand the Georgia CF Data Warehouse (GACFDW), a cloud-based high-quality computational infrastructure that is designed to support access to clinical and research data obtained from participants that have contributed to the CF Biospecimen Repository (CF-BR) and P30-supported studies. This grant will extend the GACFDW's reach to track all ~1,000 CF patients across Emory and Augusta CF Care Centers, providing comprehensive insights into both pulmonary and non-pulmonary disease progression and responses to modulator therapies.



Architecture of the Georgia CF Data Warehouse which is led by MPIs Arlene Stecenko, Nael McCarty, Rishi Kamaleswaran at Duke University, and Ryan Harris at Augusta University.

Additionally, Dr. Stecenko has received an R01 grant titled "Pulmonary and pancreatic response to cystic fibrosis modulator therapy in young children." Congratulations to Dr. Stecenko on these important achievements!

CF Biospecimen Repository (CF-BR)

Thank you to all participants and families contributing to the CF-BR. With 80% of our patients actively supporting research, your valuable samples have enabled investigators to secure NIH and CFF funding. The CF-BR now supports 53 projects and collaborates globally, advancing CF research and driving new discoveries. Your commitment is essential to this progress—thank you for making a difference.

- Dr. Arlene Stecenko, Director of the CF-BR

Research Spotlight

Clinical Trial of Trikafta Treatment in Adolescents and Adults

CF-AIR and CF@LANTA primary investigators Rachel Linnemann, MD and Eric Sorscher, MD recently published an impactful journal article which outlines the results of a recent clinical trial in people with cystic fibrosis (CF) that demonstrates significant positive outcomes, with the objective of increasing treatment access for individuals with rare genetic causes of CF.

Co-first author Dr. Rachel Linnemann and senior author Dr. Eric Sorscher, along with collaborators at the University of Alabama at Birmingham, published study results on August 26, 2024, in *The Lancet Respiratory Medicine*. The article is titled "Evaluation of elexacaftor–tezacaftor–ivacaftor treatment in individuals with cystic fibrosis and CFTR-N1303K in the USA: a prospective, multicentre, open-label, single-arm trial." This clinical trial studied treatment with elexacaftor-tezacaftor-ivacator (Trikafta) among adolescents and adults with CF who have an N1303K CFTR gene variant and a second minimal function variant, as both variants are not currently eligible for Trikafta.

The study, funded by the CF Foundation and US National Institutes of Health, enrolled 20 participants (ten male and ten female) across two sites between June 2022 and October 2023. Dr. Linnemann also presented these results alongside fellow co-first author Dr. George M. Solomon of University of Alabama at Birmingham at the CF Foundation's Therapeutics Development Network, in an April 2024 plenary session.

The team observed no significant reduction in sweat chloride after 28 days of treatment, yet they identified clinically meaningful improvements in secondary endpoints, including lung function, respiratory symptom scores, and nutritional outcomes. This trial demonstrates a new approach for testing modulator medications in rare CFTR variants with a goal of expanding access.

The Lancet Respiratory Medicine is a world-leading respiratory medicine and critical care journal with an Impact Factor of 38·7, ranking first among 54 critical care and 100 respiratory system journals globally.

Dr. Linnemann serves as the Director of the Cystic Fibrosis Care Center and Co-Director of the Pediatric Cystic Fibrosis Program at Children's and Emory, and she is also Associate Professor, Department of Pediatrics at the Emory University School of Medicine. Dr. Sorscher is a Georgia Research Alliance Eminent Scholar, the Hertz Endowed Professor in Cystic Fibrosis Research, and Professor, Department of Pediatrics at Emory University.

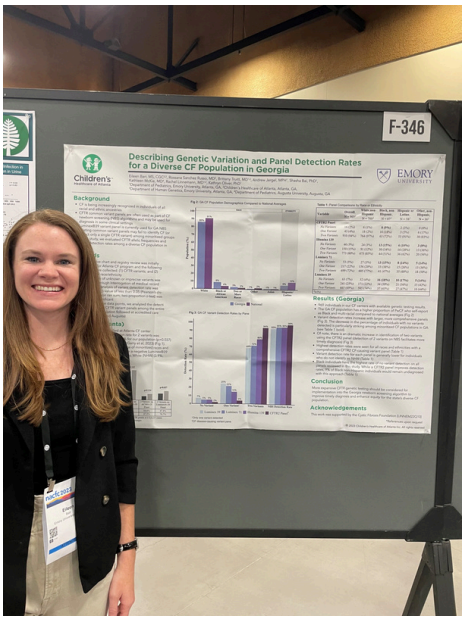
The abstract of the article can be found online here.

> ***"We are excited to share this compelling, clinical trial evidence demonstrating that individuals with the prevalent N1303K-CFTR variant exhibit substantial pulmonary improvement following treatment with elexacaftor-tezacaftor-ivacaftor. Our collaborative results with investigators in Birmingham add significantly to prior reports. These data may help pharmaceutical colleagues expand drug eligibility to include a new and important CF patient population," said Dr. Linnemann."***

Impact of Philanthropy

Flynn Family Foundation's Sustained Support for Genetic Counseling and Quality Improvement

Our work at CF-AIR and CF@LANTA is made possible through diverse funding sources, including institutional support from Children's and Emory, external grants, and generous donor contributions. These gifts are essential, allowing us to fund crucial activities that institutional budgets alone cannot cover. ***A prime example is our Genetic Counseling Program, which thrives thanks to the ongoing philanthropic support from the Patrick Flynn Memorial Foundation***.



Pictured Above: Eileen Barr, MS, Licensed Genetic Counselor, at the North American Cystic Fibrosis Conference in November 2023

The CF Genetic Counseling Program is vital to the highest quality care for the health and well-being of our patients and families.

The program:

- Offers genetic counseling at pediatric and adult CF clinics
- Improves access to genetic testing
- Provides support for newborns with CF, CRMS, and CF carriers
- Guides CF families considering IVF and preimplantation genetic diagnosis
- Counsels adolescents and adults with late CF diagnoses
- Reviews and updates missing genetic results in the CFF registry for continuity of care

Thank you!

We would like to express our gratitude and appreciation to the Flynn Family for its sustained support and commitment to the Genetic Counseling program. This program is making a significant and lasting impact on our patients and families, and it would not be possible without their continued dedication.

Impact of Philanthropy

Achievements of our Genetic Counseling Program

In addition to providing clinical care, our genetic counselor's work has included the following accomplishments:

- **Two Conference Posters/Presentations as first and second author at North American Cystic Fibrosis Conference, Phoenix, AZ, in November 2023:** Describing genetic variation and panel detection rates for a diverse cystic fibrosis center in Georgia and Evaluating Georgia's cystic fibrosis newborn screening algorithm to inform recommendations for improving quality and equity

- **Co-authored a review article in *Paediatric Respiratory Reviews* in March 2024:** Diagnostic challenges in CFTR-related metabolic syndrome: Where the guidelines fall short

- **National Involvement:** Member of the NSGC National Society of Genetic Counseling CF Special Interest Group

- **Quality Improvement Measures:**
  - CRMS: Planning and implementation of new CRMS guidelines for infants identified on newborn screening with indeterminate diagnosis
  - Infants of mother's on Trikafta: Standardize referral protocol for newborns of mothers with CF on Trikafta to refer infants to the pediatric program when needed for testing (in cases with potential false negative newborn screen)
  - Georgia Newborn Screening and Genetics Advisory Committee: Member, CF Subcommittee
  - Collaboration with Dr. Rachel Linnemann's team and the Department of Public Health/Georgia Public Health Laboratory to improve CF newborn screening:
    - New ability to add genetic testing for high-risk infants to their newborn screening by notifying the state lab of the newborn at risk (CF sibling or child to known CF carriers) and reflex to mutation testing regardless of results of first tier testing
    - New care pathway to provide genetic counseling and testing to infants with a positive newborn screen who have inconclusive sweat testing due to insufficient sweat volume. This reduces delays in diagnosis for these infants
    - New meconium ileus checkbox on the state newborn screening card to improve diagnosis in this population
    - New approval by the Georgia Newborn Screening and Genetics Advisory Committee for implementing an expanded CF DNA panel to improve equitable diagnosis, once funding is identified

Researcher Updates

Joanna Goldberg, PhD

Joanna Goldberg, PhD, Professor in the Department of Pediatrics and Microbiology & Immunology

Joanna Goldberg, PhD, was named a 2023 Fellow of the American Association for the Advancement of Science (AAAS), a prestigious lifetime honor recognizing her outstanding contributions to "pathogenic microbiology, particularly for strategies used by bacteria to cause infections in patients with cystic fibrosis." She has published over 190 original research articles in this field, and her work was recently awarded an NIH R01 grant to develop monoclonal antibody therapeutics targeting EF-Tu in Pseudomonas aeruginosa and an NIH R21 and a CFF Research Grant to study the efficacy of R-pyocins against P. aeruginosa. She and her team also have numerous productive collaborations including with local colleagues at Emory University, the University of Georgia, and Georgia Tech. They also collaborate with investigators worldwide. Dr. Goldberg is on the Board of Directors for the American Society for Microbiology and as well as an editor for numerous journals. Moreover, she has a keen interest in mentoring the next generation of scientists. See https://scholarblogs.emory.edu/goldberglab/ for additional information on the personnel and research interests in the Goldberg Lab.

Jocelyn Grunwell, MD, PhD

Dr. Jocelyn Grunwell was recently awarded an R01 grant to advance her research on pediatric acute respiratory distress syndrome (PARDS), focusing on how neutrophil cell death mechanisms impact poor clinical outcomes and linking specific PARDS endotypes with phenotypes to inform personalized treatment strategies. She was also a recipient of a Junior Faculty Focused Grant at Emory University and an NIH K23 career development award. Now on it's last year, the K23 grant has been instrumental in expanding her expertise in biostatistics, big data, and clinical trial design. With a strong background in biochemistry and molecular methods, Dr. Grunwell is establishing a comprehensive research program to uncover unique biological mechanisms in PARDS and bring precision therapies to critically ill children.

Jocelyn R. Grunwell, MD, PhD, Assistant Professor of Pediatrics in the Division of Critical Care Medicine at Emory University and Children's Healthcare of Atlanta.

Brian Vickery, MD

Brian Vickery, MD, Marcus Professor of Pediatric Immunology at Emory's School of Medicine and chief of the Division of Allergy & Immunology at Children's Healthcare of Atlanta

Dr. Vickery's groundbreaking work in recent years in food allergy led to the February 2024 approval by the Food & Drug Administration of omalizumab, also known by the brand name Xolair, for the reduction of allergic reactions — including reducing the risk of anaphylaxis — in certain adults and children one year or older with one or more food allergies. Emory University and Children's Healthcare of Atlanta were one of ten sites to take part in the first stage of the trial — known as OUtMATCH, "Omalizumab as Monotherapy and as Adjunct Therapy to Multi-Allergen OIT in Food Allergic Children and Adults" — administered nationwide through the National Institute of Allergy and Infectious Diseases-funded Consortium for Food Allergy Research. Findings were published in the New England Journal of Medicine paper titled "Omalizumab for the Treatment of Multiple Food Allergies" in February 2024, as well as in a paper in September 2024, "Omalizumab Implementation in Practice: Lessons Learned from the OUtMATCH Study."

Research Awards & Accomplishments

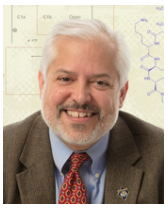
Our researchers are currently engaged in grants with national and international reach. In addition, we have been recognized throughout the scientific community for ground-breaking work in both cystic fibrosis and asthma.

Dio Kavalieratos, PhD
Member, Board of Directors, American Academy of Hospice and Palliative Medicine
R01: "Advancing Symptom Science and Management in Cystic Fibrosis: Biological, Social, and Clinical Mechanisms (PULSE – CF)"

Ben Kopp, MD, MPH
R01: "The role of CFTR during macrophage-mediated killing of bacteria"
R01: "Rescue of CF phagocyte function with CFTR modulator therapy"
CFF: "Environmental inequities and immunosenescence in CF"

Rabindra Tirouvanziam, PhD
Schinazi Family Distinguished Biomedical Chair
R01: "Extracellular Vesicle-Driven Inflammation in CF Lungs"
CFF: "Neutrophil dysfunction in CF Related Diabetes"

Jessica Alvarez, PhD, RD
Grant of Tenure in Rank, 2024
R01: "A Dietary Intervention to Improve Glucose Tolerance in Adults with Cystic Fibrosis"



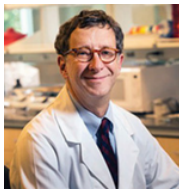
Nael McCarty, PhD
P30: "GA CF Core Center"
CFF: "Mechanisms linking mutant CFTR to dysregulated barrier function, insulin receptor function, and glucose transport"

Kimberly Dickinson, MD
CFF: "Improving Shared Decision-Making in Pediatric CF Care: Creation of a Novel Decision Aid to Guide Parents Considering G-tube Placement"

Tanicia Daley, MD, MPH
R21: "High vs Low Glycemic Index Mixed Meal Tolerance Test in Children and Adolescents with Cystic Fibrosis"

Joshua Chandler, PhD
R01: "Neutrophil Hyperexocytosis and Acid Exposure in early CF Lung Disease"

Kathryn Oliver, PhD
CFF: "Accelerating Change in Georgia's CF Newborn Screening Program to Improve Quality and Equity"



Eric Sorscher, MD
Marcus Foundation: "Improved diagnostic precision and personalized treatment for adult bronchiectasis - a mechanism-based approach"

Rachel Linnemann, MD
CFF: "Accelerating Change in Georgia's CF Newborn Screening Program to Improve Quality and Equity"
CFF: "An Open-label Trial to Evaluate the Efficacy of ETI in Subjects with Cystic Fibrosis and an N1303K Mutation Who are Not Currently Eligible for an FDA-approved Modulator"

Anne Fitzpatrick, PhD, RN, CPNP, MSCR
K24: "Mentored patient-oriented research in preschool wheezing disorders"
R01: "Phenotypes and Endotypes of Preschool Wheeze"
R01: "Symptom clusters in children with exacerbation-prone asthma"

News Around the Lab

Awards received....

Analia Vazquez Cegla, PhD received “Program Scholar of the Year” for the MSP Doctoral Program

Crystal Cobb, MD awarded CFF Second Year Clinical Fellowship and “Leann Rittenbaum Ott Fellow Award” for CF Scholars in 2024

Christian Crisan, PhD CFF Trainee Grant

Mary Ellen Fain, MD received the “Warshaw Research Award” in 2024 to study Primary Cilliary Dyskinesia

Deepali Luthra, PhD awarded a “Novo Nordisk Foundation Fellowship”

Hazel Ozuna, PhD CFF Trainee Grant

Miquéias Lopes-Pacheco, PhD CFF “Path to a Cure” Research Grant

Jiafeng (Stuart) Song received “Leann Rittenbaum Ott Fellow Award” for CF Scholars in 2023

Recent talks...

Brian Dobosh, PhD in Seoul, South Korea

JaNise Jackson, PhD at the Pediatric ECR Conference

Deepali Luthra, PhD in Copenhagen

Hazel Ozuna, PhD & Sam Durfey, PhD at the CFF Basic Research Conference in June 2024

Congratulations to CF-AIR Investigators Awarded 2023 & 2024 Pilot Grants

JaNise Jackson, PhD, with Drs. Kathryn Oliver & Yuhong Du, on "Developing novel, effective, and well-tolerated small molecule inhibitors of RPL12 that rescue disease-causing genetic variants"

Ben Kopp, MD, MPH, with Drs. Paul Dawson, Rabin Tirouvanziam & Joshua Chandler on “Immunophenotyping aerodigestive responses to new therapies in children with cystic fibrosis (CF)”

Sujit Sharma, MD, with Drs. Nael McCarty & Guiying Cui, on “Measuring real-time electrophysiological changes in an ex vivo model of human airway epithelia to identify patterns of early trans-cytosolic inflammatory injury upon respiratory viral infection”

Sohail Khoshnevis, PhD on “The epitranscriptomic regulation of Candida albicans infection in cystic fibrosis models”

Nael McCarty, PhD with 16 investigators, on “Research into the Biogeography of Inflammation-Infection-Injury and Lung Disrepair”

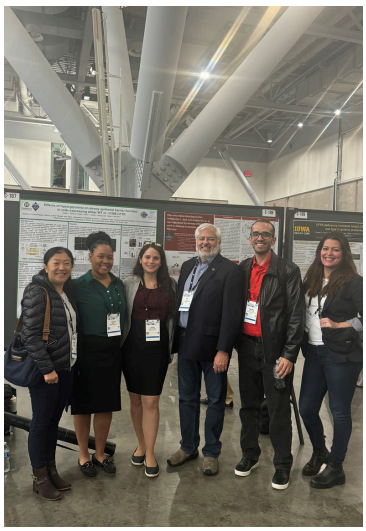
Rabin Tirouvanziam, PhD and Joshua Chandler, PhD’s SYNERGY grant with Australian colleagues on “Role of the Cytidine-Uridine Axis in Airway Macrophage Displacement by Neutrophils in Early CF Disease“

2024 NACFC

SEPTEMBER 25-28, 2024

38th Annual North American Cystic Fibrosis Conference
Boston, Massachusetts

Congratulations to our team for a successful week in Boston, with **29 platform presenters** and **over 60 posters** representing Emory, Children's & affiliates. The team was able to connect, collaborate and share insights with members of the scientific community from around the world. From recognition of great work during the plenaries (Tanicia Daley, Jessica Alvarez, Nael McCarty), to podium presentations, session chairs, and first-time poster presenters, it was an impactful conference for all CF-AIR and CF@LANTA participants.



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